



WISP3 gene

WNT1 inducible signaling pathway protein 3

Normal Function

The *WISP3* gene provides instructions for making a protein that appears to be involved in bone growth and the maintenance of cartilage, which covers and protects the ends of bones. The function of the WISP3 protein is not well understood. It is part of a family of proteins that are involved in the growth and maintenance of connective tissues, such as bone, cartilage, and blood vessels. The WISP3 protein is made in cells called chondrocytes, which produce and maintain cartilage, and is associated with the production of certain proteins that make up cartilage, but its role in their production is unclear. WISP3 may also help control signaling pathways involved in the development of cartilage and bone and may help regulate the breakdown of cartilage components.

Health Conditions Related to Genetic Changes

[juvenile idiopathic arthritis](#)

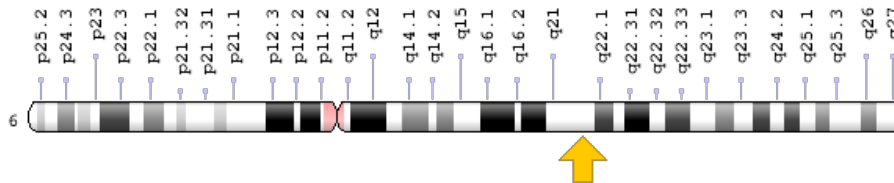
[progressive pseudorheumatoid dysplasia](#)

Mutations in the *WISP3* gene cause progressive pseudorheumatoid dysplasia (PPRD), which is a condition that causes stiffness and pain in the joints of the hands, hips, knees, and spine. The joint problems worsen over time, and movement in the joints becomes limited. Most of the mutations involved in this condition lead to production of an abnormally short WISP3 protein that is probably nonfunctional. Other mutations change single protein building blocks (amino acids) in the protein. Loss of WISP3 protein function likely disrupts normal cartilage maintenance and bone growth, leading to the joint problems in PPRD.

Chromosomal Location

Cytogenetic Location: 6q21, which is the long (q) arm of chromosome 6 at position 21

Molecular Location: base pairs 112,052,813 to 112,069,686 on chromosome 6 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CCN family member 6
- CCN6
- LIBC
- PPAC
- PPD
- WISP-3
- WISP3_HUMAN
- WNT1-inducible-signaling pathway protein 3

Additional Information & Resources

GeneReviews

- Progressive Pseudorheumatoid Dysplasia
<https://www.ncbi.nlm.nih.gov/books/NBK327267>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28WISP3%5BTIAB%5D%29+OR+%28WNT1+inducible+signaling+pathway+protein+3%5BTIAB%5D%29%29+OR+%28WNT1-inducible-signaling+pathway+protein+3%5BTIAB%5D%29+OR+%28WISP-3%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- WNT1-INDUCIBLE SIGNALING PATHWAY PROTEIN 3
<http://omim.org/entry/603400>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
<http://atlasgeneticsoncology.org/Genes/WISP3ID469ch6q22.html>
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=WISP3%5Bgene%5D>
- HGNC Gene Family: CYR61/CTGF/NOV matricellular proteins
<http://www.genenames.org/cgi-bin/genefamilies/set/1046>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=12771
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/8838>
- UniProt
<http://www.uniprot.org/uniprot/O95389>

Sources for This Summary

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